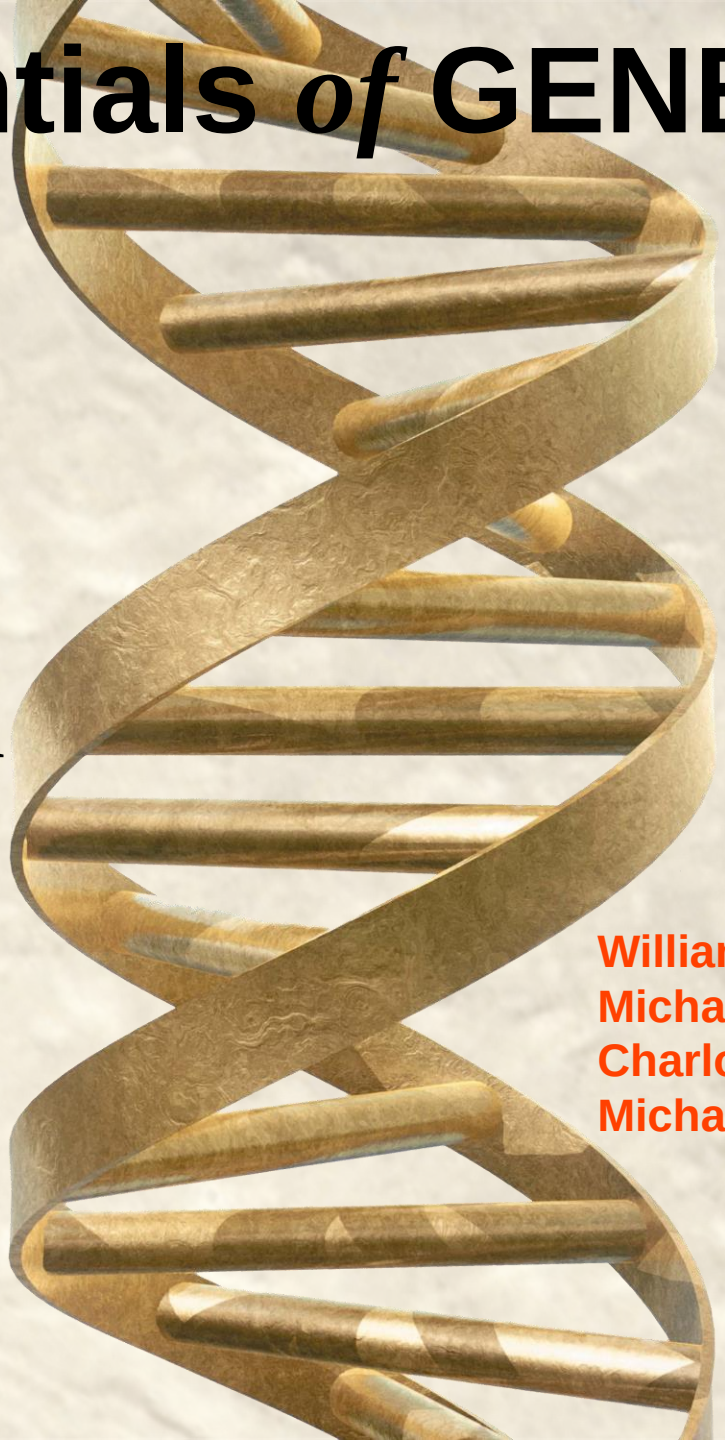
Essentials *of* GENETICS

Eighth Edition



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Introduction to Genetics

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Chapter 1 Introduction

- In December 1998, Iceland granted deCode Genetics, a biotechnology company, a license to create and operate a database drawn from medical records of all its 270,000 residents until 2012.
- A combination of medical-genealogical information forms a powerful resource available exclusively to deCode.
- This is a real example of the increasingly complex interaction of genetics and society.

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Chapter 1 Introduction

- Similar large-scale projects have been generated in Great Britain, Estonia, Latvia, the Kingdom of Tonga, and other countries.
- In the United States, smaller-scale programs involving tens of thousands of individuals are underway.
- These databases will be used to search for genes that control human diseases.
- It is important to address the ethical questions surrounding an emerging technology as the information is gained.

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1.1 Genetics Has a Rich and Interesting History

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Section 1.1

- Archaeological evidence documents thousands of years of domestication and cultivation of plants by artificial selection.
  - 8000 to 1000 B.C.: domestication of horses, camels, oxen and wolves; maize, wheat, rice, and date palm
- The Hippocratic School of Medicine (500–400 B.C.) and Aristotle (384–400 B.C.) attempted to explain heredity using the concept of active “humors” and vital heat.

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Section 1.1

- 300 B.C.–A.D. 1600: Few significant ideas were put forward to explain heredity.
- 1600–1850: The Dawn of Modern Biology
  - William Harvey (1600s): Theory of **Epigenesis**
    - An organism develops from a fertilized egg through developmental events, transforming the egg into an adult.
  - Schleiden and Schwann (1830): The Cell Theory
    - All organisms are composed of cells derived from preexisting ones.

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Section 1.1

- Louis Pasteur disproved the idea of **spontaneous generation.**
  - Creation of living organisms from nonliving components
- Charles Darwin's travels on the HMS *Beagle* provided him geological, geographical, and biological observations that helped formulate his theory of **natural selection.**

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Section 1.1

- Darwin published his ideas on evolutionary theory in *The Origin of Species* (1859).
- Existing species arose from other ancestral species by descent with modification.
- Natural selection was the driving force for evolutionary change.
- Evolution was independently proposed by Alfred Russel Wallace.
- Darwin had no understanding of the mechanism involved.
- Mendel (1866) offered an explanation using peas.

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1.2 Genetics Progressed from Mendel to DNA in Less Than a Century

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Section 1.2

- Mendel worked with peas and used quantitative data to show that traits are passed from parents to offspring in predictable ways.
  - Each trait is controlled by a pair of genes that separate during gamete formation.
- Mendel published his findings (1866) offering a general model of how traits are passed, but his work was largely unknown.
- Mendel's work was "rediscovered" around 1900 and forms the foundation of genetics.

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Section 1.2

- Advances in microscopy have identified chromosomes (**Figure 1-2**) and establish that most eukaryotes are diploid (2n, two sets of chromosomes).
  - Human diploid number: 46 (**Figure 1-3**)
- Eukaryotic cells undergo two types of cell division:
  - **Mitosis:** Two resulting daughter cells receive a **diploid** set of chromosomes (**Figure 1-4**).
  - **Meiosis:** Resulting cells (gametes) receive only half the number of chromosomes (**haploid,** n).

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Section 1.2

- The chromosomal theory of inheritance (Sutton and Boveri) states that inherited traits are controlled by genes residing on chromosomes.
- Genes are transmitted through gametes.
- This transmission maintains genetic continuity from generation to generation.

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Section 1.2

- Alternate forms of a gene are called alleles.
  - That is, alleles for various eye colors.
- Variations in genes/alleles (DNA sequences) are the result of mutations.
- Mutations are the source of genetic variation.
- The set of alleles for a given trait is called the genotype.
- The expression of the genotype produces an observable trait or phenotype.

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Section 1.2

- DNA (nucleic acid), not protein, is the carrier of genetic information
  - Research of Avery, MacLeod, and McCarty: 1944

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1.3 Discovery of the Double Helix Launched the Era of Molecular Genetics

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Section 1.3

- The structure of DNA was described by Watson and Crick (1953).
- DNA is an antiparallel, double-stranded helix.
  - Monomer: nucleotide
    - Sugar bonded to a phosphate and four bases
      - Adenine, cytosine, guanine, and thymine
  - These nucleotides form A=T and G=C complementary base pairing across the helix (**Figure 1-7**).

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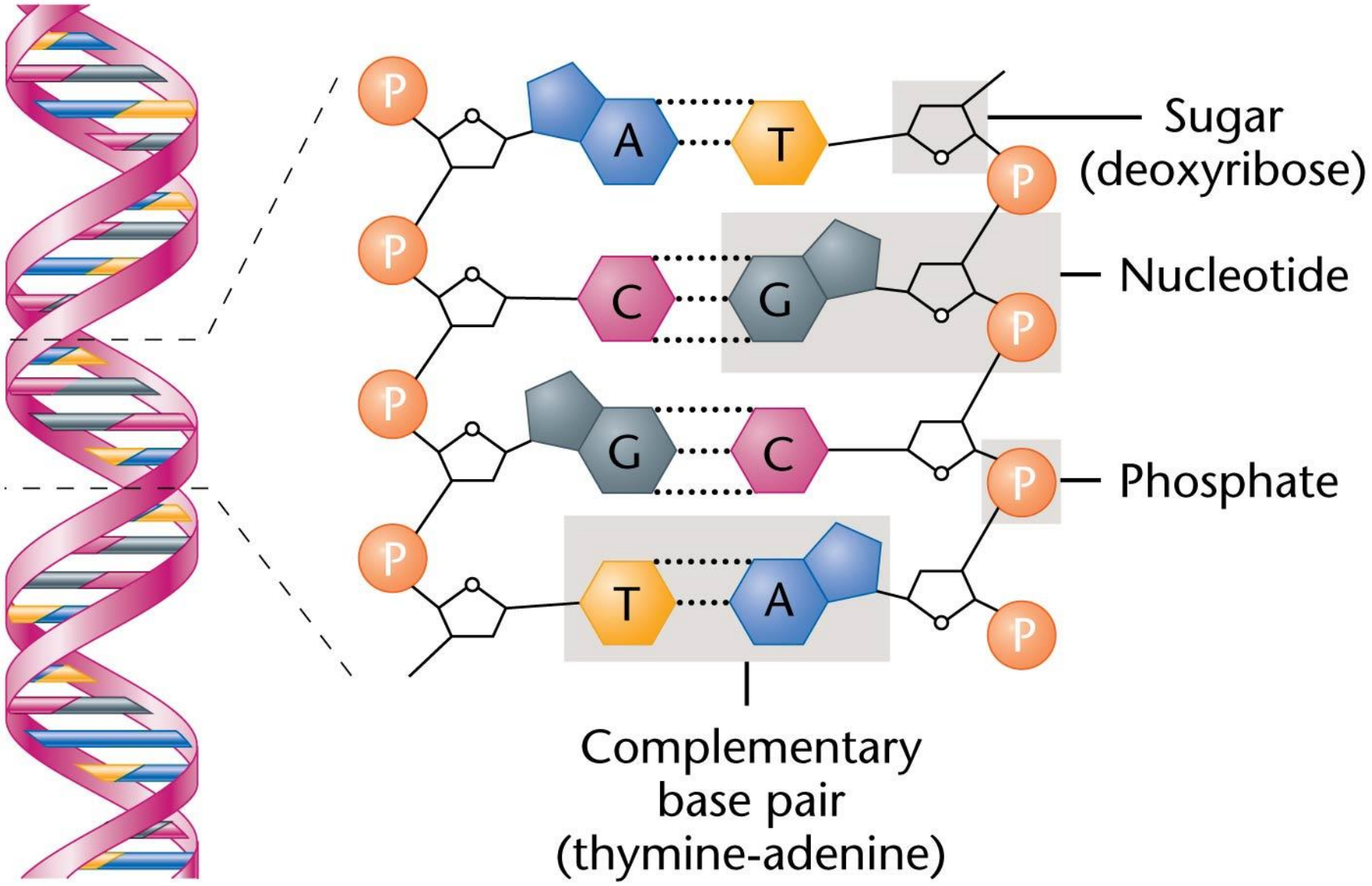
Figure 1-7



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Section 1.3

- RNA (nucleic acid) is similar to DNA, except that:
  - it is usually single-stranded.
  - it has adenine, cytosine, and guanine but has uracil (U) in place of thymine (T).
  - the sugar in RNA nucleotides is ribose instead of deoxyribose.

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Section 1.3

- **Transcription** is the process by which the information on a DNA strand is transcribed into a messenger RNA (mRNA).
  - Transcription occurs in the nucleus.
- The mRNA moves into the cytoplasm where it binds to a ribosome.
- The information in the mRNA is translated into a protein (**translation**).
- This is known as the central dogma of genetics (**Figure 1-8**).

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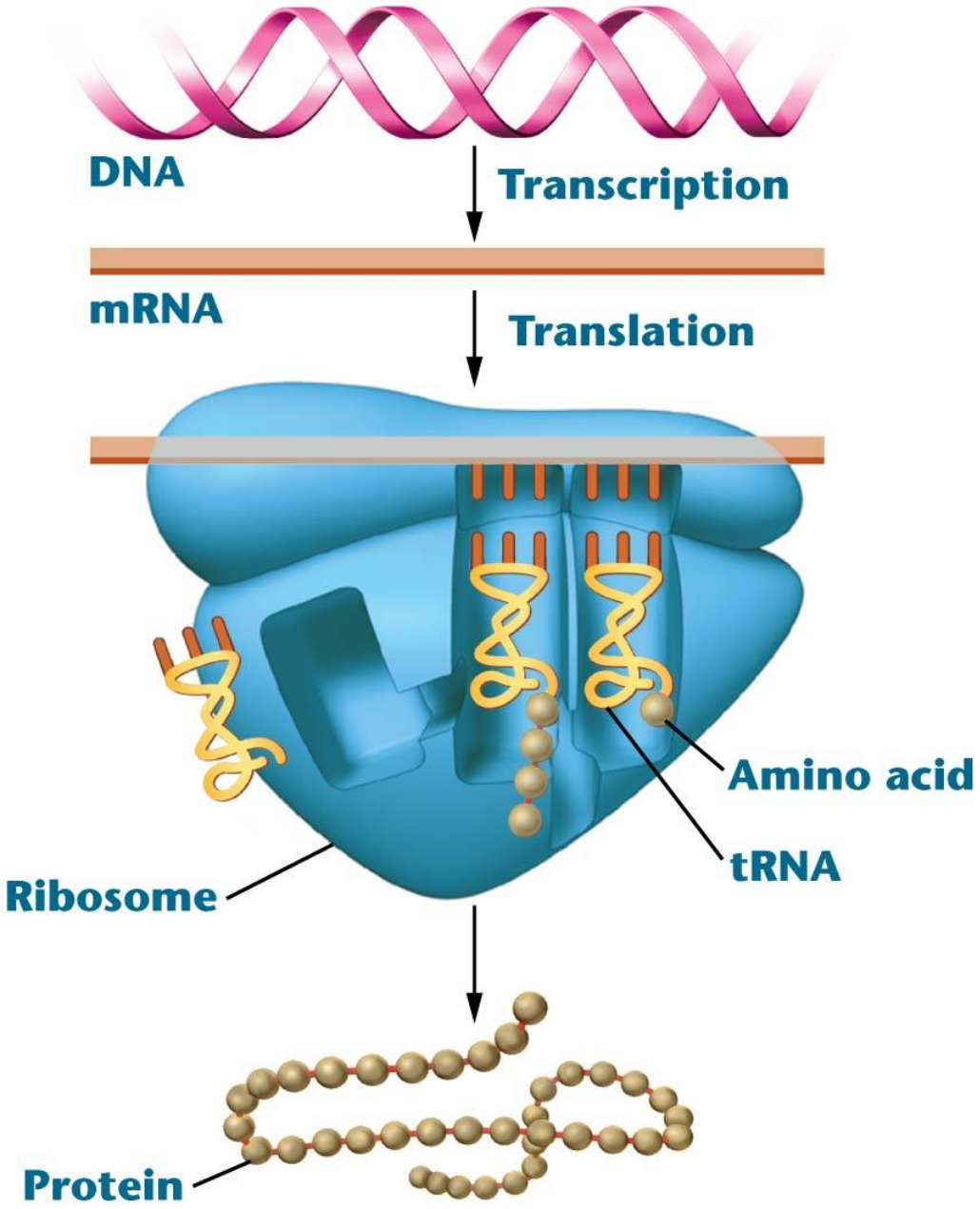
Figure 1-8



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Section 1.3

- The genetic code consists of triplet nucleotides present in mRNA.
- Each triplet (codon) encodes for insertion of a specific amino acid into a growing protein chain.
  - There are 20 different amino acids commonly found in proteins.

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Section 1.3

- Once a protein is made, its action or location in a cell plays a role in producing a phenotype.
  - Enzymes are the largest category of proteins.
  - Other proteins include hemoglobin, insulin, connective tissue, actin, and myosin.

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Section 1.3

- Mutations altering a gene may modify, alter, or even eliminate the protein's usual function and cause an altered phenotype.
  - Sickle-cell anemia results from a mutation in the gene encoding B-globin, resulting in an amino acid substitution (**Figure 1-9**).
  - Sickle-shaped red blood cells are deformed and fragile and break easily, leading to a whole series of physical and physiological problems (**Figure 1-10**).

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1.4 Development of Recombinant DNA Technology Began the Era of Cloning

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Section 1.4

- In the 1970s researchers discovered **restriction enzymes** in bacteria that cut viral DNA at specific sites.
- With the use of vectors, restriction enzymes have allowed the advent of recombinant DNA and cloning.
- Recombinant technology has given rise to the biotechnology industry, which is a major contributor to the U.S. economy.

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1.5 The Impact of Biotechnology Is Continually Expanding

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Section 1.5

- Biotechnology has been used for the genetic modification of crop plants for:
  - increased herbicide, insect, and viral resistance.
    - 85% of U.S. corn plants
    - 95% of U.S. soybean plants
  - nutritional enhancement.
- Some genetically altered traits in crop plants are shown in **Table 1.1**.

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Section 1.5

- Biotechnology has also been used for the genetic modification of animals (**Figure 1-11**).
  - Makes possible the production of even dozens or hundreds of offspring with desirable trait
  - Transgenic animals are used to synthesize therapeutic proteins.
    - Anticlotting protein from milk of transgenic goats
    - Clinical trials underway to treat several diseases, including emphysema

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TABLE 1.1 Some Genetically Altered Traits in Crop Plants

Herbicide Resistance	Corn, soybeans, rice, cotton, sugarbeets, canola
Insect Resistance	Corn, cotton, potato
Virus Resistance	Potato, yellow squash, papaya
Nutritional Enhancement	Golden rice
Altered Oil Content	Soybeans, canola
Delayed Ripening	Tomato

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Section 1.5

- Gene therapy and genetic testing have had profound impact on the diagnosis of genetic diseases.
- The molecular basis for hundreds of genetic disorders is known (**Figure 1-12**).
- Biotechnologically derived prenatal diagnosis of “carriers” is currently available for more than 100 inherited disorders.

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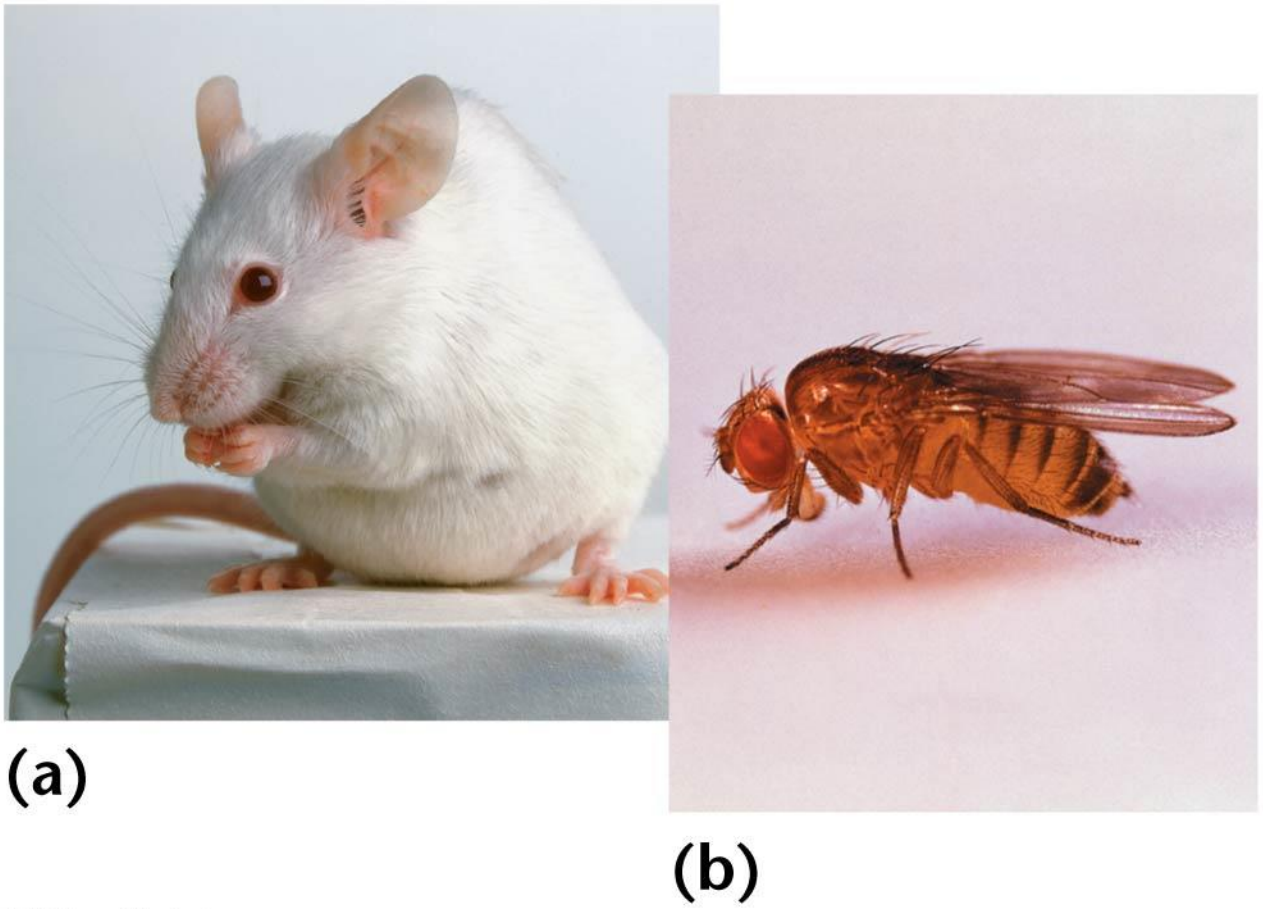
Figure 1-13



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1.6 Genomics, Proteomics, and Bioinformatics Are New and Expanding Fields

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Section 1.6

- **Genomics** analyzes genome sequences to study the structure, function, and evolution of genes and genomes.
- **Proteomics** identifies a set of proteins present in cells under a given set of conditions and studies their function and interactions.
- **Bioinformatics** develops hardware and software for processing nucleotide and protein data.

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1.7 Genetic Studies Rely on the Use of Model Organisms

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Section 1.7

- Model organisms for genetic study meet the following criteria:
  - Easy to grow
  - Short life cycle
  - Produce many offspring
  - Genetic analysis straightforward

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Section 1.7

- All life has a common origin, and genes with similar functions in different organisms are similar in structure and DNA sequence.
- The genetics of model organisms can be applied to humans for understanding and treating human diseases.

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Section 1.7

- By transferring genes between species, scientists have developed models of human diseases in organisms ranging from bacteria to fungi, plants, and animals (**Table 1.2**).
- The gene transfer approach is being used to study many human neurological disorders including
  - Huntington Disease
  - Alzheimer's Disease

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TABLE 1.2 Model Organisms Used to Study Some Human Diseases

Organism	Human Diseases
E. coli	Colon cancer and other cancers
S. cerevisiae	Cancer, Werner syndrome
D. melanogaster	Disorders of the nervous system, cancer
C. elegans	Diabetes
D. rerio	Cardiovascular disease
M. musculus	Lesch–Nyhan disease, cystic fibrosis, fragile-X syndrome, and many other diseases

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1.8 We Live in the Age of Genetics

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Section 1.8

- The field of genetics started with Mendel's presentation of his research on peas in 1865.
- Genetics has rapidly developed to include genomics today (**Figure 1-15**).
- Many genetics-related Nobel prizes have been awarded.
- Society is faced with a host of sensitive genetics-related issues, including prenatal testing, genetic discrimination, ownership of genes, and access to/safety of gene therapy and private ownership.

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